ASSESSING THE NEEDS FOR A SYSTEM OF GENETIC SERVICES

STATE OF HAWAII

PACIFIC SOUTHWEST REGIONAL GENETICS NETWORK

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ASSESSING THE NEEDS FOR A SYSTEM OF GENETIC SERVICES FOR THE STATE OF HAWAII

I. INTRODUCTION AND PURPOSE

As the Human Genome Project nears completion in the first decade of the 21st century, knowledge flowing from it will begin to have a major impact on medicine and other sciences, industry, agriculture, law and the environment. The stage will be set for an Age of Genetics that could rival the Industrial Revolution in its impact on society.

—Time, Beyond the Year 2000: What to Expect in the New Millennium, Fall 1992.

The importance of genetics on the future health and well-being of the people in our communities can be far-reaching as evidenced in the following examples:

> At the turn of the century, the infant mortality rate in the United States was approximately 150 per 1,000 live births. It was estimated that about five of those 150 deaths could have had a genetic-related cause. Since that time the infant mortality rate has declined dramatically primarily due to improvements in medical and health services such as prenatal care, nutrition, environmental conditions, and the control of infectious diseases. While the number of infant deaths due to environmental and infectious causes have decreased; the relative proportion of deaths due to genetic-related disorders have increased so that now nearly one-third of all infant deaths are due to genetic-related causes.
>
> It is now recognized that there are three major factors that can have an impact on the health of all individuals: the environment in which the individual lives, the lifestyle and behavioral choices that the individual makes, and the genetic makeup that the individual inherits. Of these three factors, the greatest advances have been made in the field of genetics.

As medical technology expands and improves, the importance and impact of genetics in preventing, alleviating and curing some of the major causes of morbidity and premature mortality due to genetic defects becomes more of a reality. In order to channel the efforts in the field of genetics to improve the effectiveness of the service delivery system, a model for a comprehensive and integrated system is proposed.

The purpose of the document is to:

(1) Present a conceptual framework for the development and implementation of an administrative infrastructure to assure that a comprehensive system with a full continuum of genetic services can be provided to all residents in the State of Hawaii.

(2) Describe the existing program structure and the services that are currently being provided.

(3) Make recommendations for improvements in the administrative structure and the service delivery system.

II. DESCRIPTION OF THE STATE OF HAWAII

The Hawaiian Island chain stretches for 1,523 miles northwest to southeast, from the tiny island known as Kure Atoll down to Cape Kumukahi, the easternmost point of the "Big Island" (the Island of Hawaii). Seven of the eight most southerly islands are the inhabited portions of the state. The entire State of Hawaii has a total land mass of 6,425 square miles.

The City and County of Honolulu, on the island of Oahu, is the center of government and business for the State of Hawaii. Although the smallest of the four counties geographically, it has about 75.4% of the State's population. For most purposes, the City and County of Honolulu is defined as the entire Island of Oahu, an area of 594 square land miles. The resident population of the County of Honolulu as of April 1, 1990 was estimated at 836,231 persons. This total includes active members of the armed forces and their dependents, which comprise approximately 15% of the total population on Oahu.

Hawaii County, encompassing the Island of Hawaii, is the state's largest county in size. With an area of 4,034 square land miles, the Island of Hawaii is almost twice the combined size of all the other islands in the State. The estimated resident population of the County in 1990 was 120,317 persons, which is 10.8% of the State's total. There are 216 statute miles between Honolulu and Hilo, the County seat.

Kauai County includes the islands of Kauai and Niihau, the two northernmost major inhabited islands of the Hawaiian chain. Niihau is a privately owned, entirely rural island having a total population of 230 persons in 1990 and is only accessible to the general public at uninhabited sites. Kauai is the State's third largest and least populous of the four counties. The Island of Kauai is 549 square land miles in area and Niihau is 70 square miles. The estimated population for the island of Kauai in 1990 was 51,177 persons and represents 4.6% of the total state population. The distance between Lihue, the County seat, and Honolulu is 102 statute miles.

Maui County is the second largest county in the State of Hawaii and includes four major islands--Maui, Lanai, Kahoolawe, and a major portion of Molokai. The total land area for Maui County is 1,162 square miles, of which Maui makes up 729 square miles; Molokai, 261 square miles; Lanai, 140 square miles; and uninhabited Kahoolawe being 45 square miles. In 1990, the estimated county resident population was 100,504 persons or 9.1% of the total state population. Of this total population, 91,361 persons reside on the Island of Maui, 6,717 individuals on Molokai Island (including 130 residents of Kalaupapa in Kalawao County) and 2,426 residents on Lanai. There are 101 statute miles between Honolulu and Kahului, the County seat of Maui.

DESCRIPTION OF THE GENERAL POPULATION

The results of the 1990 census reported a Statewide resident population of 1,108,229; well over the 1980 census count of 946,691 residents. These census results include members of the armed forces and their dependents, a group making up 10.4 percent of the resident population.

The State of Hawaii has many unique characteristics, among which is the fact that the State of Hawaii is an island state with its residents populating seven separate and distinct islands. Hawaii is also unique in the makeup of the ethnic variability and the lack of a racial majority. Table 1 lists the predominant self-reported ethnic groups and the corresponding percentages based on the 1990 census data. Though no ethnic group constitutes a majority, the Caucasians are the largest group with 33.4% of the population followed by the Japanese with 22.3%, Filipinos at 15.2% and Native Hawaiians at 12.5%.

DISTRIBUTION OF ETHNICITY

ETHNIC GROUP	PERCENT
Caucasian	33.4
Japanese	22.3
Native Hawaiian	12.5
Filipino	15.2
Chinese	6.2
Black	2.5
Korean	2.2
Vietnamese	0.5
Samoan	1.4
Other/Unknown	3.9

Source: Census Data, 1990

Table 1

The population of the state is relatively young with a median age of 32.6 years, mostly male (51%) and racially diversified. In 1990, there were 258,285 women of child bearing ages between the ages of 15 and 44 years. During this same period for this population of women, there were 26,834 recorded reproductive events which resulted in 20,438 live births, 1,689 standard fetal deaths (20 or more weeks of gestation), and 4,707 intentional terminations of pregnancy.

Of the total live births in 1990 in the state, 15,380 (75.3%) were born to women who were residents of Honolulu County, 2,228 (10.9%) to women of Hawaii County, 945 (4.6%) births to Kauai residents and 1,885 (9.2%) to Maui County residents, (Maui - 1,714 births, Molokai - 129 births, and Lanai - 42 births).

Based on the estimated total population, the birth rates for the State of Hawaii, and the estimated probability of risk for a genetic disorder; the number of individuals in each of the risk groups can be predicted as follows: 30,000 couples who are in the reproductive age groups, 25,000 reproductive events, 20,000 newborns, 2,000 infants with "birth defects", 8,000 children and adults with genetic disorders, and an unknown number of individuals at risk for genetic problems (Figure 1).

III. OVERALL CONCEPT OF A SYSTEM OF GENETIC SERVICES

Because of the complexity of the situation, no one agency, public or private, in the State of Hawaii has all the capability nor the resources to address the problems alone. Therefore, it will take a system approach and the coordinated and cooperative efforts of many organizations, agencies, and programs. However, government agencies have a specific role in assuring appropriate public health measures. Based on the Institute of Medicine's, *The Future of Public Health* (1988), the role of government is:

1. Assessment. The committee recommended that every public health agency regularly and systematically collect, assemble, analyze, and make available information on the health of the community, including statistics on health status, community health needs, epidemiologic and other studies of health problems.

2. Policy development. The committee recommended that every public health agency exercise its responsibility to serve the public interest in the development of comprehensive public health policies by promoting use of the scientific knowledge base in decision-making about public health and by leading in developing public health policy. Agencies must take a strategic approach, developed on the basis of a positive appreciation for the democratic political process.

3. Assurance. The committee recommended that public health agencies assure their constituents that services necessary to achieve agreed upon goals are provided, either by encouraging actions by other entities (private or public sector), by requiring such action through regulation, or by providing services directly. It is also recommended that each public health agency involve key policymakers and the general public in determining a set of high-priority personal and community-wide health services that governments will guarantee to every member of the community. This guarantee should include subsidization or direct provision of high-priority personal health services for those unable to afford them.

To make a significant impact on the prevention and control of morbidity and premature mortality due to genetic-related causes in the State of Hawaii, an overall system will need to be developed and implemented that will assure that the public

agency address the recommendations in a rational approach. This system must organize our existing resources into an integrated service delivery system that will promote efficiency, avoid unnecessary duplication, improve access to care, and enhance the quality of services in order to respond to the needs of the citizens of Hawaii.

There are four major subsystems in the overall system: a) the Administrative subsystem, b) the Service Delivery subsystem, c) the Data subsystem, and d) the Quality Assurance subsystem. (Figure 2)

A. THE ADMINISTRATIVE SUBSYSTEM

The Administrative subsystem is the central component of the overall system and is the base of the infrastructure that is required to assure that all the other components in the overall system are developed, implemented, and integrated. The Health Department's Family Health Services Division and Children with Special Health Needs Branch will be the agency responsible for assuring the development and implementation of the Administrative subsystem. The primary activities included in the Administrative subsystem include:

PLANNING ON A STATEWIDE BASIS - The planning process is a set of activities that include assessing the community needs, identifying specific health problems or issues, defining strategies and developing implementation, developing and allocating resources for service delivery, and evaluation of effectiveness.

FORMULATING POLICIES - In response to the addressing the problems related to the morbidity and premature mortality of persons affected with genetic-related disorders, the Health Department will need to formulate and promulgate policy statements that will recommend the principles, standards, priorities and initiatives to be used by public officials, decision makers, as well as service providers.

ASSURING COORDINATION AND COMMUNITY RELATIONS - The intent of coordination and maintaining community relations is to establish on-going and open communications and encourage participation in order to provide the opportunity for the Health Department to work with the community agencies in being responsive to the community needs. The Health Department will be responsible for providing the leadership in coordinating all activities related to the prevention, treatment, and rehabilitation of genetic-related disorders.

PROVIDING TECHNICAL ASSISTANCE - In accepting the leadership role, the Health Department should be responsible for assuring a level of professional standards in this area and must provide the technical

assistance and professional guidance that will be required by personnel at the service delivery levels. To fulfill this responsibility, Health Department program staff must maintain the expertise in the latest developments, the findings in current research, current political trends, and new models of service delivery. In addition, the staff will also need to maintain expertise in process skills, teaching skills, and administrative consultation to assist in the development and maintenance of an effective system.

B. THE SERVICE DELIVERY SUBSYSTEM

In order to make an impact on the incidence and consequences of genetic related conditions, we must organize our resources into coordinated efforts with common understandings, common goals, strategies and outcomes.

It will require the cooperative efforts of the programs in both the private and public sectors to assure a full continuum of high quality services to include prevention and outreach, screening and testing, diagnosis, counseling, treatment, support services, and case management (Figure 2).

C. THE DATA SUBSYSTEM

High quality data and information are essential for the planning, development and evaluation efforts in any human service program. In order for statewide data to be available, the Health Department will be responsible for developing a data system that will accomplish the following:

1. Serve as a centralized database for the purposes of identifying clients and for demographic surveillance.
2. Serve as an interactive system that will allow the tracking of service provision.
3. Serve as a mechanism for evaluation of service utilization, client outcomes, and system functions.

CENTRAL STATEWIDE DATA BASE

The development and implementation of a centralized and integrated data base begins with a health record for each patient. This data is generated at the service delivery program level and specific demographic and diagnostic data is conveyed to a central data entry point. This statewide data base should serve as the central registration system that will be able to provide demographic information related to all individuals receiving genetic services in the system. There are many issues that will need to be examined, assessed and decided upon. Some of the issues are:

1. CONFIDENTIALITY MUST BE ABSOLUTELY ASSURED. As with any patient/client specific identification system, confidentiality at all levels must be maintained to comply with applicable federal and state requirements. Some methods of assuring confidentiality are: (1) modifications in software that allow limited access to certain portions of the data base; (2) establish a numeric coding system for identification of and access to patient files together with a physically separate master listing of names and numeric codes.

2. Referrals and transmittal of data to the central data base. With the Children with Special Health Needs - Genetics Program responsible for establishing, implementing and maintaining the central data base, the main issue becomes the development of mechanisms for data transmittal from a variety of community based programs which are the primary sources of data. Some of the other issues are: (1) incentives that can be offered to providers of services to make the initial referrals and updating the data for the central data base; (2) types of training and education that will be required for personnel submitting data; (3) definitions, procedures and forms for submitting data.

3. Feedback of information to programs/agencies submitting data. Mechanisms and formats for reporting of information retrieved from the central data base will need to be developed.

4. Data base management. Procedures will need to be developed for the management of the daily operations of the data base as well as procedures for the periodic updating and maintenance.

SERVICE TRACKING

Tracking is a process of continuous monitoring and assessment of the services received by an individual and/or family to meet their identified needs. The tracking system should be designed for the following outcomes: (1) Patients are not "lost" in the system of service delivery; (2) timely provision of services is assured; (3) needed services are identified based on a comprehensive evaluation; and (4) the provision of the services is appropriately monitored. There are many issues that will need to be assessed as the tracking system is designed, developed and implemented. Some of these issues are:

1. Comprehensive assessment and individual plans. Mechanisms, policies and procedures will need to be developed for conducting comprehensive assessments for the identification of needs and methods of documenting

those needs into an individual plan.

2. Data generation and submission. With the service delivery system designed as a community based system, service provision is accomplished by a variety of programs, agencies, facilities and individual providers. Policies, procedures, methods of data collection and transmittal, forms for collecting data, among others will have to be developed and implemented.

3. Daily operations and maintenance of the tracking system. Procedures will need to be developed for the compiling of data, the editing and validating of data and data entry.

EVALUATION

Evaluation is an inherent part of everyday living as well as part of all program activities. Evaluation is part of the cyclical process of planning and assesses the effectiveness and the degree of success in achieving predetermined objectives. Therefore, one of the important and early steps in the development of an evaluation process is the establishment of indicators:

1. System indicators-measure the effectiveness of the system by being able to identify individuals in need of service, to assure that the service delivery system is responsive to the needs of the individual.

2. Service indicators-measure the utilization of services and reflect the degree to which services are available and accessible to the population.

3. Outcome (status) indicators-measure the health (outcome) status of individuals within the service delivery system. The health status is the accumulative interaction of lifestyle, health practices, attitudes and availability and accessibility to services.

D. THE QUALITY ASSURANCE SUBSYSTEM

Quality assurance is a comprehensive program of activities that are designed and implemented to enhance the quality of care being provided through the ongoing objective assessment of certain aspects of patient care. In reviewing the historical development of quality assurance efforts, professional accountability stands out as a predominant force in influencing and shaping the activities of quality assurance programs. The intent of quality assurance is the correction of problems identified through this process. The basic elements of the process of quality assurance includes: (1) optimal standards of care, (2) a comparison between the actual care provided against the standards, (3) the identification of deficiencies, (4) corrective action, and (5) the assessment of the effects of the corrective action.

Quality assurance as a comprehensive concept can be implemented by any one of the four different organizational entities (funding agency, regulatory agency, professional organization, service provider agency); and depending on the organizational entity conducting the quality assurance, the intent, the approach, the methodology, and the expected outcomes will be different.

There are several organizational entities at various levels that use various mechanisms with which quality assurance can be implemented.

> FUNDING AGENCIES - use monitoring as a means to evaluate the organizational structure, performance, and effectiveness of a service agency.
>
> REGULATORY AGENCIES - use licensing to ensure safe and competent practice of individuals; and accredit or certify service agencies for quality care.
>
> PROFESSIONS - use peer review to ensure professional competency in providing services.
>
> SERVICE PROVIDER AGENCIES - use audits to evaluate the fiscal operations, performance evaluations to assess employees, and program evaluations of outcome measures to assess the effectiveness of the agency.

Each of the organizational entities has a different intent and a different approach to quality assurance; however, the mechanisms of implementing quality assurance are complementary and may be reciprocal.

In recent times, the process of establishing professional standards has become institutionalized through professional organizations, licensing or accrediting bodies, and legislation; however, we must not lose sight of the fact that the health care provider must be a key participant in the development and evolution of standards. Based on this concept of assuring professional accountability in proving quality services, it is proposed that the philosophy of promoting high quality services through education and support be the impetus in designing quality assurance programs for the field of genetics. The implementation of this philosophy would include: (1) developing standards in a cooperative effort among the service providers, professional organizations and the public agency; (2) developing methods of monitoring and evaluation with the intent of identifying areas for professional education, training and technical assistance; (3) development mechanisms for the provision of technical education, training and technical assistance in order to improve the quality of services provided.

IV. DESCRIPTION AND ANALYSIS OF PROGRAMS

The description and analysis of each of the categories of programs and services will be discussed in the following manner. First, a model spectrum of services will be described for each of the category of services — what should be in place. Immediately following will be a description of the array of services in each of the categories that are available — what is actually in place in Hawaii. When appropriate, a third section will describe the identified need for services.

A. PRIMARY PREVENTION

1. A MODEL SPECTRUM OF PREVENTION SERVICES

Genetic diseases affect all economic, social, age, racial, ethnic and religious groups. For the State of Hawaii, it is estimated that every year there are over 25,000 reproductive events, 20,000 live births, 2,000 infants with "birth defects", 8,000 children and adults with genetic disorders, and many more persons who are at risk for genetic disorders or have genetic susceptibility. It is to these individuals at risk that genetic disorders has its effects — at least 50 percent of spontaneous abortions are caused by chromosomal abnormalities, approximately one-third of all infant deaths are due to genetic-related causes, and nearly one-third of all hospital bed days in children between the ages of one to four years are because of genetically determined or influenced disorders.

Rather than trying to address the problems that are the results of genetic disorders, a more effective strategy for addressing these problems is with primary prevention through education. Primary prevention can be described as a set of educational activities that are for the general public or targeted for specific audiences and designed to reduce the incidence of genetic disorders by providing individuals and families with adequate information to make the appropriate decisions related to future reproductive outcomes.

The majority of preventive programs and activities involve raising the awareness, informational level, and the involvement of the individual, family and community. These types of programs are based on the assumption that individuals in the community are not sufficiently aware of the risks and the problems associated with genetic disorders; nor the alternatives available for families with or at risk for genetic conditions. These activities are generally targeted for the general public and take the form of print media campaigns, telephone information lines, public service announcements, community events such as health fairs, speakers at community meetings, community councils and coalitions.

2. AVAILABLE PREVENTION SERVICES IN HAWAII

The availability of preventive educational programs and activities for the general public and high risk families in Hawaii is limited. Table 2 lists the primary agencies and programs that provide the array of preventive services. All of these agencies are based on the Island of Oahu and therefore the majority of services are provided on Oahu. Some of the services are provided on the Neighbor Islands through the local chapters and programs on each of the islands, or with persons traveling from Oahu.

ACTIVITY	PROGRAM/AGENCY
Public Service Announcements	Maternal and Child Health - Health Department March of Dimes, Chapter of the Pacific
Informational brochures	Children with Special Health Needs - Health Department Health Promotion and Education - Health Department Maternal and Child Health - Health Department Medical Genetic Services Fetal Diagnostic Center - Kapiolani Medical Centers March of Dimes, Chapter of the Pacific Muscular Dystrophy Association Hemophilia Foundation National Neurofibromatosis Foundation - Hawaii Chapter
Community events	Children with Special Health Needs - Health Department Medical Genetic Services March of Dimes, Chapter of the Pacific
Videotapes	Medical Genetic Services March of Dimes, Chapter of the Pacific Fetal Diagnostic Center - Kapiolani Medical Centers
Speakers for community	Children with Special Health Needs - Health Department Medical Genetic Services Fetal Diagnostic Center - Kapiolani Medical Centers March of Dimes, Chapter of the Pacific
Information telephone lines	Tel-Med - Hawaii Medical Services Association Call-a-nurse - Kapiolani Medical Centers Teratology Information Services - Kapiolani Medical Centers ASK-2000
Classes	Medical Genetic Services Fetal Diagnostic Center- Kapiolani Medical Centers March of Dimes, Chapter of the Pacific Queen's Medical Center

Table 2. Programs Providing Preventive Genetic-Related Services, State of Hawaii

3. THE IDENTIFIED NEEDS FOR A SYSTEM

There are a multitude of agencies, programs and services that provide awareness and education of issues related to genetics and genetic services. However, despite this array of educational services, the impact at the community level is not effective because of the lack of coordination and collaboration among these efforts. In order to be effective, a comprehensive and coordinated preventive education effort should be planned and implemented.

The Health Department's Children with Special Health Needs Branch must assume the leadership for the planning of a multi-agency collaborative effort to develop and implement a coordinated multi-media genetics awareness and educational campaign. These efforts should target the general public to increase the awareness of the impact of genetics on pregnancy outcomes, to improve the knowledge base of the importance of genetics in sub-populations at risk for genetic disorders, and to facilitate access to genetic screening, diagnosis and treatment services.

B. SCREENING PROGRAMS

1. A CONCEPTUAL MODEL OF SCREENING PROGRAMS

The Commission on Chronic Illness (1957) defined screening as the presumptive identification of unrecognized disease or defect by the application of tests, examinations, or other procedures which can be applied rapidly. Cohen (1978) more specifically defines screening programs for genetic disorders..."primarily as an activity in preventive medicine aimed at detecting individuals in whom it is likely that a specific hereditary disease will develop for which preventive or therapeutic measures are available". Further developments have extended the definition to include individuals or couples who are at risk for having affected offsprings as the results of a particular genetic constitution or abnormality. The National Academy of Sciences' Committee for the Study of Inborn Errors of Metabolism (1975) defines genetic screening as the search in a population for persons possessing certain genotypes that: (1) Are associated with disease or predispose an individual to disease. These persons are identified to permit monitoring for early signs of a disease in those who are genetically predisposed to that disease, or are identified for curative or supportive treatment. (2) May lead to disease in the individual's descendants. These Individuals include persons who are or may be carriers for disorders. They are identified so that genetic counseling may be provided regarding their reproductive options and risks. (3) May produce other variations not known to be associated with disease. These Individuals may be identified for research purposes such as determining frequency in a population (Natowicz, 1991).

Before any genetic screening program is implemented, consideration must be

given to five issues:

(1) The rationale for the development of the program must be considered in terms of the importance and desirability of screening. The importance of a genetic condition means that it is prevalent in the population or that it is associated with very serious problems for a subset of the population.
(2) The quality of the screening test and the quality of its implementation must be evaluated. A screening test should be both sensitive and specific, as well as rapid, safe, inexpensive, and reproducible.
(3) Screening for a genetic condition must be both appropriate and feasible. Appropriate mechanisms must be set in place so that the individuals to be tested will have access to testing centers, and that the centers have adequate resources and facilities to perform the test well and to provide adequate follow-up. Also the information elicited at the screening should be pertinent for the family.
(4) A cost-benefit analysis should indicate that is economically effective to establish and run the screening programs.
(5) Any effective genetic testing program must have an appropriate and effective system for processing, storing, and following-up the information that is generated.

In designing and implementing genetic screening programs, potential hazards must be recognized and guarded against. These hazards include: (a) the possibility of missing individuals by using inaccurate screening tests; (b) the risk of increasing community anxiety; (c) the communication of positive results too late to prevent the manifestation of the disease; (d) the lack of benefit to the patient who is diagnosed; (e) poor or inadequate counseling; and (f) excess economic burden to the state, family, or individual.

There are several different approaches to genetic screening; and may be classified on the basis of the types of subjects identified. The following are some of the currently recognized types of screening programs being offered: (a) screening of asymptomatic heterozygote individuals in the pre-conceptual period, (b) screening in the prenatal period, and (c) screening of newborns.

The screening of asymptomatic individuals for many different types of health conditions has become more common as prevention and health supervision become a larger part of health care. The capability of screening to find asymptomatic heterozygote individuals for genetic conditions through new laboratory techniques has seen a rapid growth. However, screening programs for asymptomatic individuals have not been as effective as newborn or prenatal screening. Until the potential harm that might result from the confusion, stigmatization, discrimination, and inappropriate use of the data is balanced with

the benefits of testing, the implementation of mass screening programs for asymptomatic individuals will be difficult.

Prenatal screening and diagnosis refers to all the technologies currently in use to determine the physical and physiologic condition of a fetus before birth. These technologies include amniocentesis, chorionic villus sampling, tests on maternal blood samples, and ultrasound screening. Prenatal screening and diagnosis in conjunction with appropriate and quality genetic counseling will provide prospective parents at risk with information to make an informed choice related to the pregnancy.

Screening in the newborn period was first introduced in 1961 and since that time has become an important and integral part of care of the newborn. The system relies primarily on primary care practitioners which makes them an integral part of any screening program. The practitioners are responsible for parent education, the collection and handling of samples, and prompt action on any abnormal results. It is the role of the state agency in collaboration with the practitioners to assure that every newborn is screened, that abnormal results are tracked and all affected infants provided with quality diagnostic and treatment services.

2. AVAILABLE SCREENING SERVICES IN HAWAII

a. Pre-Conceptual / Heterozygote Screening

There are no legislative mandated programs in the State of Hawaii that address the issues or provides the services related to pre-conceptual and heterozygote screening.

The Comprehensive Hereditary Anemia Program for Hawaii had been in operation between 1985 and 1992 under the auspices of the Medical Genetics Services of the University of Hawaii, John A. Burns School of Medicine. This program provided thalassemia and G6PD screening for high risk minority populations.

The objectives of the program were to detect heterozygote individuals at risk for lethal or handicapping inherited anemias in Southeast Asians in Hawaii through the following: (a) health professional and lay education, (b) family recruitment, (c) improvement in diagnostic techniques and data interpretation, (d) understanding of results by clients, and (e) assuring effective utilization of health resources. Other objectives included assisting affected patients by providing preventive treatment to minimize the consequences of the disease; identify couples at risk and offer them genetic counseling and fetal testing; improve the provision of health services for the at-risk population; and optimize screening protocols for heterozygote individuals by efficient use of available techniques, and development of new screening techniques.

With the termination of federal funding for this demonstration project, community screening services had been terminated; however, pre-conceptual diagnostic testing services continues with funding from the Kapiolani Medical Centers.

b. Prenatal Screening

On April 15, 1988, the Legislature of the State of Hawaii enacted Act 23 which amended Chapter 321 of the Hawaii Revised Statutes and added Section 331 that allowed the Health Department to adopt rules to "ensure that all pregnant women in the state are offered appropriate information, quality testing, diagnostic services, and follow-up services concerning neural tube defects and other disorders amenable to prenatal diagnosis". Hawaii Revised Statutes, Chapter 321-331 further stipulated that the Health Department may (1) provide educational resources to all women in the state before and early in pregnancy about the availability of prenatal tests, counseling, information on the benefits, risks and limitations of prenatal tests; (2) make available prenatal screening and diagnostic tests to all pregnant women in the state who choose to be screened; (3) specify the diseases which may be screened for; (4) determine screening and diagnostic test methodologies; (5) establish laboratory quality control standards; (6) provide technical assistance to laboratories, hospitals, physicians, and other health care providers; (7) maintain confidential registry and collect appropriate statistical data for research and evaluation; (8) collect fees for program services; and (9) maintain confidentiality of records.

The Prenatal Screening Program is administered by the Health Department's Children With Special Health Needs Branch and has a personnel complement of one full time Program Coordinator, one full time Tracking Nurse, one half time Laboratory Technician and one full time and one half time clerical staff.

The Prenatal Screening Program seeks to assure that all pregnant women who present for prenatal care before 20 weeks gestation are offered appropriate information, quality testing and follow-up services concerning neural tube defects and other disorders detectable by prenatal diagnosis.

During 1992, community health care providers and commercial laboratories began offering "triple screening" (combination of MSAFP, maternal serum human chorionic gonadotropin and maternal serum unconjugated estriol). The Prenatal Screening Program is currently assessing the impact of the triple screening on the activities of the program. Other activities of the program include upgrading the data collection system between the Prenatal Screening Program and the service providers and participating laboratories. Linkages between the Prenatal Screening Program, the Health Department's Office of Health Status Monitoring and the Birth

Defects Monitoring Program are being developed to improve the monitoring of test results.

c. Newborn Screening

Hawaii's Newborn Screening Program began in 1965 with the passage of Chapter 331-1, Hawaii Revised Statutes that mandated screening of all newborns for phenylketonuria (PKU). In 1983, screening for congenital hypothyroidism was added. In 1986 the Hawaii State Legislature repealed Chapter 331-1, HRS and amended Chapter 321 and added section 291 which allowed the Health Department to "specify diseases to be screened for in newborn infants and methods to be employed to best prevent mortality and morbidity within the population of the state". Chapter 321-291 further mandated the Health Department to adopt the rules necessary rules to implement this section to include (1) administration of newborn screening tests; (2) quality and cost control of screening tests; (3) keeping of records and related data; (4) reporting of positive tests, (5) guidelines for care, treatment, and follow up of infants with positive test results; (6) informing parents about the purposes of the tests; and (7) maintaining the confidentiality of affected families.

The Newborn Screening Program is administered by the Health Department's Children With Special Health Needs Branch and has a personnel complement of one full time Program Coordinator, one full time Tracking Nurse, one half time Laboratory Technician, and one full time clerical staff.

The objectives of the Newborn Screening Program are to prevent and ameliorate the effects of handicapping conditions that are identified through the administration of newborn screening and diagnostic testing. The program is responsible for administering the state's newborn metabolic screening statute which requires that all newborn infants be screened and tested for phenylketonuria and congenital hypothyroidism. The activities to fulfill these responsibilities include the planning, development and implementation of the screening program, tracking and follow-up of infants to assure satisfactory testing, assuring that all infants with the specified diseases are detected and program with appropriate and timely treatment.

Financial assistance for comprehensive diagnostic and treatment services is available for income-eligible families of children with PKU and congenital hypothyroidism. Staff coordinating services are available to all families regardless of income eligibility for services.

Expansion of newborn screening is taking place with the following activities: (1) On January 1, 1993, the Newborn Screening Program and the Kapiolani Medical Centers began conducting a pilot study on the feasibility of screening newborns for

congenital adrenal hyperplasia. (2) On May 1, 1993, the Tripler Army Medical Center began screening newborns for sickle cell and other hemoglobinopathies.

3. THE IDENTIFIED NEEDS IN SCREENING SERVICES

1. There is a need to assess the feasibility and effectiveness of establishing a consistent funding base for the operation of a screening program to identify heterozygote individuals pre-conceptually for hereditary anemias.

2. Currently the Newborn Screening Program is routinely screening all newborns for only two conditions - PKU and Congenital Hypothyroidism. Pilot studies are under way to test the feasibility of adding congenital adrenal hyperplasia and hemoglobinopathies. According to the Council of Regional Networks for Genetic Services (CORN) 1990 Newborn Screening Report, many states are screening newborns for several other conditions. A total of 42 states screen for hemoglobinopathies, 38 states screen for galactosemia, 22 states screen for Maple Syrup Urine Disease, 21 screen for Homocystinuria, 14 screen for Biotinidase deficiency, and 8 states screen for adrenal hyperplasia. By using the criteria presented above, the Health Department's Newborn Screening Program needs to assess the feasibility and effectiveness of adding other newborn screening tests.

C. MEDICAL DIAGNOSIS AND FOLLOW-UP

1. A MODEL OF MEDICAL DIAGNOSTIC / FOLLOW-UP SERVICES

Advances in genetics brought on by the recognition of the importance of genetic factors in common diseases in adulthood, new screening techniques for carrier detection and presymptomatic diagnosis, and improved therapy for genetic disorders have created an awareness of and an increased demand for clinical genetic services. These services which focus on the diagnosis, clinical management, and genetic counseling have been supported by a variety of federal, state, foundation, and private sector funds. With improved and increased funding, clinical genetics have evolved from its beginnings as a primarily research oriented service provided at academic medical centers to one that is now widely available through centers in the community. Most comprehensive genetic centers provide a variety of services that focus on diagnosis, management and follow-up, and counseling. Those programs that are medical-center based provide inpatient consultative services, outpatient services, and consultation in subspecialty clinics.

The genetics evaluation consists of accumulating the data and family information necessary to reach the correct diagnosis in the proband. The diagnostic evaluation usually consists of the family history, history of the pregnancy, delivery and

neonatal period, the physical examination, appropriate laboratory investigations, and family investigations.

After a diagnosis has been established, the medical facts and the interpretation of the facts concerning the diagnosis can be discussed with the family. This process, genetic counseling, has been defined by the American Society of Human Genetics as "...a communication process which...involves an attempt of one or more appropriately trained persons to help the individual or the family to (1) comprehend the medical facts, including the diagnosis, the probable course of the disorder and the available management; (2) appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives; (3) understand the options for dealing with the risk of recurrence; (4) choose the course of action which seems appropriate to them in view of their risk and their family goals and act in accordance with that decision; and (5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder."

Appropriate follow-up is a crucial component of genetic services. Follow-up visits may be used to assure understanding of the information related to the diagnosis and the risks of recurrence provided earlier in the evaluation. Visits may also be used to provide recommended treatments and medical management for family and patients.

Most comprehensive genetic centers are located in large medical centers with the availability of a comprehensive multidisciplinary array of health care professionals with expertise in their respective disciplines. In these centers, the core providers are clinical geneticists (physicians with subspecialty training and board certification in clinical genetics) and genetic counselors (health professionals with graduate-degree level training and board certification in genetic counseling). In addition to these core providers, genetic services should also involve medical social workers and nurses who are able to provide psychosocial support, crisis intervention, family follow-up, and referrals to community resources. Nutritionists and dietitians are also important members of the service teams. In some situations, persons who may assist families in bereavement may also be included on the team.

2 AVAILABLE DIAGNOSTIC AND FOLLOW-UP SERVICES IN HAWAII

a. Diagnostic evaluation and management

1. Clinical genetic specialists

Dr. Y.E. Hsia, the founder and Director of Medical Genetic Services, came to Hawaii from Yale University in 1977. For many years he was the only clinical geneticist in the state and has provided medical genetic services on a contractual

basis for the Health Department's Children with Special Health Needs Branch. He has been the recipient of 36 genetics-related research and service grants; is internationally recognized for research on hereditary hemoglobinopathies and von Hipple-Landau disease; and has served as visiting professor to Australia, China, Indonesia, and Thailand. He currently holds a tenured professorship with the University of Hawaii John Burns School of Medicine.

The objectives of the Medical Genetic Services program include: (1) To offer medical genetic services to the people of Hawaii. (2) To provide student and professional continuing education in medical genetics. (3) To promote genetic awareness in the health community. (4) To improve preventive care standards for genetic illnesses. (5) To collect, process, and apply new information about genetic diseases. (6) To integrate activities in medical genetics in Hawaii, nationally and internationally.

Since 1977, the Medical Genetic Services has provided over 7,500 genetic inquiries, family evaluations, and consultations to physicians and other health professionals. Direct clinical services have been provided on a statewide basis with outreach clinics on all of the neighbor islands. These direct services include patient evaluations, special diagnostic testing, family carrier screening, and genetic counseling. The Medical Genetic Services has also provided teratogen information services on a statewide basis.

Increased genetic awareness and education has been achieved by the Medical Genetic Services through a variety of methods. The program staff have provided lectures and seminars to students at the University of Hawaii in the undergraduate school, medical school, and school of public health for many years. The staff has also participated in continuing education activities for physicians and other health professionals throughout the state. Awareness of genetic services has been accomplished by community educational activities, publicity in the medical literature and mass media, and collaborative educational activities with community volunteer organizations.

Dr. Greigh Hirata, Director of Reproductive Genetics at Kapiolani Medical Centers, has been the staff perinatologist and clinical geneticist with the Fetal Diagnostic Center since 1990. He trained at the University of Southern California and Cedars-Sinai Medical Center in Los Angeles. He is board certified in medical genetics and perinatology. He currently holds a position Assistant Professor of Maternal and Fetal Medicine, Department of OB/Gyn, University of Hawaii John Burns School of Medicine.

The Fetal Diagnostic Center at the Kapiolani Medical Centers offers specialized diagnostic services that includes amniocentesis, ultrasound, percutaneous

umbilical blood sampling, and chorionic villus sampling. The Fetal Diagnostic Center is the only obstetrical clinic to provide a comprehensive and integrated array of services for pregnant women who are at high risk. Services may include perinatal consultation, comprehensive treatment and management of high risk pregnancies, prenatal diagnosis, antepartum fetal testing, fetal surgery, medical genetics, patient education, and referrals. The center is staff by four perinatologists, with one of the perinatologist also a clinical geneticist, three genetic counselors, four nurses and support staff.

Dr. Dwight Yim is a pediatrician with the Kaiser Permanente Foundation Hospital and provides genetic services for the patients enrolled in the Kaiser health maintenance organization's service delivery system. He sees patients upon referral on an individual appointment basis and provides the diagnostic and clinical management services required. There are no genetic counselors or specific genetic clinics.

Because of the complex medical, social, and health needs of individuals with genetic disorders and their families, other private providers in the community are involved in the care of these patients and their families. These providers include perinatologists, obstetricians, pediatricians, neurologists, endocrinologists, hematologists, public health nurses, nutritionists, and social workers. It is the combined efforts of all of these clinical specialists that assures the comprehensive services required by these patients.

2. Laboratories

Genetic-related laboratory services are provided by the private sector and by the Tripler Army Medical Center. There are five laboratories in the state that are approved to provide newborn metabolic screening services (PKU and hypothyroidism). These include the Clinical Labs of Hawaii which is based in Hilo on the island of Hawaii. The other laboratories are on the island of Oahu and include Diagnostic Laboratory Services, Kaiser Reference Laboratory, Kapiolani Medical Center for Women and Children Laboratory, and Tripler Army Medical Center Laboratory. Newborn screening services are available through these laboratories at all of the facilities that provide maternity and delivery services.

There are six approved laboratories to provide prenatal maternal serum alpha fetoprotein (MSAFP) screening services in the State of Hawaii. The privately-owned laboratories include Clinical Laboratories of Hawaii (Hilo), Diagnostic Laboratory Services, Kaiser Reference Laboratory, Kapiolani Medical Centers Laboratory, and Accupath/Smithkline Beecham Laboratory. The Tripler Army Medical Center Laboratory is supported by the Federal Government and limits services to military personnel.

There are three genetics-related specialty laboratory services in the state: (1) Kapiolani Medical Centers, Molecular and Clinical Cytogenetics Laboratory, (2) Mid-Pacific Genetics, Inc., and (3) the Honolulu Police Department. In addition to these laboratories located in the state are several mainland laboratories that provide services to private community based providers - however, discussion will be limited to laboratories in the state.

The Kapiolani Medical Centers' Molecular and Clinical Cytogenetics Laboratory is headed by Dr. Timothy Donlon who received his doctorate degree in the Department of Medical Genetics at the Oregon Health Sciences University. After receiving his doctorate degree, he completed a research fellowship at the Children's Hospital in Boston. In 1986, he established the Molecular and Clinical Cytogenetics Laboratory at Stanford University. He is the chairperson for the committee on the Genetic Constitution of Human Chromosome #15, which is part of the Genome Project. Dr. Donlon is Board Certified in Cytogenetics, Molecular Genetics, and Medical Genetics.

The Molecular and Clinical Cytogenetics Laboratory performs cytogenetic analysis of amniotic fluid specimens, cancer genetic analysis which requires specialized culturing conditions for specific cell types, high-resolution analysis to increase the number of chromosome bands for microdeletion (continuous gene) syndromes, fragile X analysis and breakage analysis for Fanconi Anemia, ataxia telangiectasia and Bloom Syndrome. The DNA laboratory has the capability of performing molecular testing for the fragile X syndrome and the thalassemias. The laboratory also serves as a national referral service for the detection of abnormalities associated with the Prader-Willi/Angelman syndromes.

The Mid-Pacific Genetics, Inc. is under the direction of Dr. Mark Bogart who has had extensive experience in all aspects of clinical cytogenetics. He has served as the Division of Medical Genetics Assistant Direct at the University of California at San Diego and is currently the chairman of the Pacific Southwest Regional Genetics Network's (PSRGN) Quality Assurance Committee.

The Mid-Pacific Genetics, Inc. is a Honolulu based clinical cytogenetics laboratory that provides complete cytogenetic services including: chromosome analysis of amniotic fluid, chorionic villus samples, bone marrow and blood for hematologic disorders, blood for congenital and reproductive disorders, autopsy tissue and products of conception. In addition to standard chromosome analysis procedures, the laboratory offers testing for fragile X syndrome, high resolution banding and fluorescent in-situ hybridization.

The Honolulu Police Department operates a forensic DNA laboratory which is in the process of compiling a data base for the purposes of DNA profiling. The Honolulu

Police Department is authorized to collect blood and saliva samples from offenders convicted on homicides and sexual crimes. It is anticipated that the police department will be able to analyze DNA found at a crime scene and perform comparisons of DNA in the data base.

b. Follow-up management

1. Genetic counselors

Genetic counseling is available from three full time counselors and one half-time counselor. All the genetic counselors are at the Kapiolani Medical Centers and provide services through the Fetal Diagnostic Center and the Medical Genetic Services programs. Individual counselors have specialty areas such as high risk prenatal and pediatric genetic counseling, preconceptual and adult genetic counseling, and teratogen counseling.

2. Genetic-related clinical services

Clinical services for a variety of genetic-related conditions are provided by the public as well as the private health care sector. The following is a list of available clinics.

CLINIC	LOCATION	COUNSELOR	FUNDING
Cardiac	Kapiolani Medical Center and NI	None *	DOH
Craniofacial	UH Medical School	None *	DOH
Hemophilia	Kapiolani Medical Centers (KMC)	None *	Foundation
Metabolic	Medical Genetic Services	Yes	DOH
Myelodysplasia	Shriner's Hospital	None *	Shriner's DOH KMC
Muscular Dystrophy	Rehabilitation Hospital of the Pacific	None *	Foundation
Neurology	Kapiolani Medical Center and NI	None *	KMC
Orthopedic	Shriner's Hospital	Yes	DOH
Sickle Cell F/U	Tripler Army Medical Center (TAMC)	Unknown	TAMC
Thalassemia	Medical Genetic Services	Yes	KMC

DOH = Hawaii Department of Health
NI = Neighbor Islands
* Referred to Medical Genetic Services for genetic counselor services

3. THE IDENTIFIED NEEDS IN MEDICAL AND FOLLOW-UP SERVICES

The majority of clinical genetic services in the State of Hawaii has been centered primarily around prenatal, newborn, and pediatric services. The availability of all

types of genetic services vary widely across the state. The Kapiolani Medical Centers has been instrumental in assuring that medical services are available; however, in a centralized system of care at the medical center.

There is a need to examine alternative strategies in developing plans for expanding the availability and access to clinical genetic services. Some of the questions that will need to be analyzed include: (a) Should a centralized service delivery system be continued for should the focus be changed to a community-based system? (b) With limited and decreased funding for clinical services, how can third party reimbursements be assured to expand service provision within the context of health care reform? (c) How can genetic services be integrated into the primary health care service delivery system?

There is a need to explore the feasibility of consolidating the laboratory functions for the prenatal and newborn testing into one or two central laboratories for the purpose of maintaining the quality of test results.

D. SUPPORT SERVICES

1. A MODEL OF SUPPORT SERVICES

Families with a member affected with a disability require many resources and support to adequately care for that family member. First, parents need information about the condition - the diagnosis, how did it happen, what will the impact be on the future. One of the benefits of information is to decrease the sense of isolation. Another important resource is other parents who have had the same experience, ideally through an organized parent support group. The parents are able to not only provide the support but also can help to find appropriate and successful models for coping with the new situation.

2. AVAILABLE SUPPORT SERVICES IN HAWAII

a. Family support groups

There are several family support groups and services available on the Island of Oahu, however, few services are available on the neighbor islands. The following are some of the local family support services that are available:

(1) Alzheimer's Association, Caregiver Support Group
(2) Bereavement Support Group through the Kapiolani Medical Centers' chaplains
(3) Down Syndrome Family Support Group, Down Syndrome Congress - Hawaii

(4) Hemophilia Foundation
(5) Muscular Dystrophy Foundation
(6) Myelodysplasia Family Support Group, Kapiolani Medical Centers
(7) PKU Family Support Group, Medical Genetic Services
(8) Transfusion-Dependent Support Group, Medical Genetic Services
(9) Neurofibromatosis Support Group, National Neurofibromatosis Foundation
(10) CF Family Information and Support Group, Cystic Fibrosis Foundation
(11) Epilepsy Foundation of Hawaii

b. Agency Support

(1) Muscular Dystrophy Association, founded in 1950, is..."a national voluntary health agency - a dedicated partnership between scientists and concerned citizens aimed at conquering forty specific neuromuscular diseases..." The organization provides funds for research, patient services and clinical care, and professional and public education. There is a local chapter of the MDA which conducts a clinic at the Rehabilitation Hospital of the Pacific. The MDA is funded through the Jerry Lewis telethon.

(2) Hemophilia Foundation is a national non-profit organization with chapters in 50 states across the U.S. The Hawaii Chapter of the Hemophilia Foundation provides information, materials, advocacy (especially on insurance issues), assistance in accessing medical care, psychosocial support services, and financial assistance. A clinic for patients with hemophilia is conducted monthly at the Kapiolani Medical Centers. The Hemophilia Foundation is funded by Aloha United Way, and federal and state contract grants.

(3) March of Dimes Birth Defects Foundation is a national non-profit organization with a mission to prevent birth defects. The Chapter of the Pacific is the local chapter and has divisions on the island of Oahu and each of the major neighbor islands - Hawaii, Kauai, and Maui. The March of Dimes provides education to the general public, to health professionals and to employees at worksites. The MOD also provides funds for each of the neighbor islands for educational and small project grants. The MOD is dependent upon fundraising events for its funds.

3. THE IDENTIFIED NEEDS FOR SUPPORT SERVICES

The best resource for parents may be other parents who have had the same experience, ideally through an organized parent support group. The need for this type of support is best identified by affected individuals and families and developed through "grass roots" efforts. The health professionals and public health administrators may be a resource to these support groups through providing

guidance and advice.

V. DESCRIPTION OF ADMINISTRATIVE COMPONENTS

A. GENETIC COORDINATOR - HAWAII DEPARTMENT OF HEALTH

The goal of the Health Department's Children with Special Health Needs Branch is to prevent and ameliorate the effects of handicapping conditions in children with special health needs through early identification, intervention and treatment by assuring a comprehensive family-centered, community-based, coordinated system of services. A coordinated system of genetic services is a critical component of a comprehensive statewide system of services for children with special health needs. To achieve the goal, the branch has created in it's organization the Hawaii State Genetics Coordinator position with the overall responsibility of planning, developing, evaluating, and coordinating genetic or genetic-related services within the state.

B. KAPIOLANI MEDICAL CENTERS

The Kapiolani Medical Centers has created the position of Executive Director, Kapiolani Medical Centers Genetics Group to develop, coordinate and implement genetic activities, programs and services within the organization. The primary responsibilities of the Executive Director are' to coordinate, expand and improve genetic services within the Kapiolani Health Care System (KHCS); to assure that quality services are provided by members of the Genetics Group; to seek grants for clinical genetic research; to foster collaboration between KHCS and other genetics-related community service agencies; and to maintain communication with key participants in KHCS and the statewide genetics community through media, marketing, and public relations.

C. GENETIC ADVISORY COMMITTEES

1. NEWBORN SCREENING ADVISORY COMMITTEE-HEALTH DEPARTMENT

The purpose of the Health Department's Newborn Screening Advisory Committee is to advise the Department regarding the technical and administrative aspects of newborn screening; to provide support, guidance and feedback about newborn screening services; to provide a means of public accountability for the program; to discuss issues relevant to newborn screening including immediate problems and long range plans; to disseminate information about the program; to improve communication and coordination among the Department, health care providers, laboratories, hospitals, and the community; and to assist with advocacy for newborn screening services.

The chair of the Newborn Screening Advisory Committee is the Chief of the Children with Special Health Needs Branch. The committee is composed of individuals from all segments of the newborn screening system representing consumers, birthing facilities, laboratories, primary care physicians, endocrine and metabolic specialists, and other primary care providers. The committee meets at least twice a year and more frequently if necessary.

2. PRENATAL SCREENING ADVISORY COMMITTEE-HEALTH DEPARTMENT

The purpose of the Prenatal Screening Advisory Committee is to provide advice and feedback regarding new tests, laboratory guidelines, and education related to prenatal screening.

3. BIRTH DEFECTS MONITORING PROGRAM ADVISORY COMMITTEE

The Birth Defects Monitoring Program Advisory Committee was initially convened in 1988 after the program was established in response to a recommendation to the Governor by a community-based coalition to establish an active birth defects and adverse reproductive outcome surveillance system for the state. The responsibilities of the advisory committee were to advise the Health Department, program staff and consultants regarding the overall design and administration of the program. Members of the committee were asked to provide scientific guidance in the selection of guidelines for case identification, record abstraction, data storage and analysis, confidentiality issues, and research protocols.

The committee is comprised on 16 health professionals and administrators from both the private and public sectors. Representation on the committee is from the Health Department, University of Hawaii, School of Public Health, Cancer Research Center, Kapiolani Medical Centers - Perinatology, Neonatology, and Pediatric Departments, Queen's Medical Center, Kaiser Medical Center, Medical Genetic Services, March of Dimes, and Pacific Southwest Regional Genetics Network.

4. PACIFIC SOUTHWEST REGIONAL GENETICS NETWORK - HAWAII

The Pacific Southwest Regional Genetics Network (PSRGN) - Hawaii Genetics Committee was established in October 1990 to act as an advisory committee to the Principal Investigator of the PSRGN. The purpose of the committee is to provide support, guidance and feedback about the status of genetic services in Hawaii; plan long term genetic services; disseminate information about genetic services to the community, and improve communication and interaction among key individuals involved in the development and implementation of genetic services in the state.

The Committee is composed of nine individuals from all segments of the community representing consumers; Health Department representatives; providers

of genetic services; providers of health care services; University of Hawaii, Genetics Department; and Kapiolani Medical Centers.

D. DATA SYSTEMS

Within the state of Hawaii, a multitude of public and private agencies support a variety of manual and automated databases and data systems. The following are descriptions of the data systems:

1. Newborn Screening Program (Children with Special Health Needs Branch, Health Department). The program receives normal and abnormal laboratory test results for phenylketonuria (PKU) and congenital hypothyroidism (CH) and enters the results into a data base at the program site. The infants who have abnormal results are followed by the program until a final diagnosis is made and treatment is initiated.

2. Prenatal Screening Program (Children with Special Health Needs Branch, Health Department). The program receives normal and abnormal maternal serum alpha fetoprotein (MSAFP) test results on a voluntary basis submitted by participating laboratories. The abnormal results are entered into a data base at the program site. Data are now being transmitted via modem from the laboratories to the program.

3. Hawaii Tumor Registry (Hawaii Health Registries, Cancer Research Center of Hawaii) was established in 1960 by the Hawaii Medical Association (HMA), the Health Department (DOH), and the Hawaii Pacific Division of the American Cancer Society. The Hawaii Tumor Registry is jointly operated by the Cancer Research Center, HMA, and DOH under a contractual agreement with the National Cancer Institute's Surveillance, Epidemiology and End Results (SEER) Program. The registry contains records of all cancer patients diagnosed in Hawaii, provides complete cancer reporting for the entire state and serves as the basis for all epidemiologic cancer research and cancer control activities in Hawaii, as well as nationally. The registry is unique in that it contains data on racially diverse groups (such as Hawaiians, Japanese, Chinese, Filipino, Koreans, Samoans) that have been used to ascertain ethnic variations in cancer incidence and survival.

4. The Hawaii Birth Defects Monitoring Program (Hawaii Health Registries, Cancer Research Center of Hawaii), established in 1988, is administered through the Cancer Research Center of Hawaii on a contractual agreement with the Health Department. The program collects data at all the birthing facilities throughout the state through on-site review and abstracting of

medical records. Over 125 demographic and diagnostic data elements are collected on each infant or mother who meet program eligibility requirements. The data system includes fetal death greater than 20 weeks of gestation and therapeutic abortions performed as a result of abnormal prenatal diagnosis. The program monitors the occurrence of over 1,000 different types of birth defects. With this information, the program is able to monitor the trends and changes in the incidence of birth defects and provide an information base for developing public awareness and education programs for the prevention of birth defects. The information can also be used to determine future demands for services in order for health planners to plan and develop appropriate services and prevention strategies.

5. Cancer Research Center of Hawaii, established in 1971, has operated as an independent institute within the University of Hawaii. Organizationally, the center has four operational divisions: Molecular Oncology, Clinical Trails, Epidemiology, and Population Registries and Database which includes the Hawaii Tumor Registry and the Hawaii Birth Defects Monitoring Program. The Cancer Research Center's goal is to discover how cancer can be prevented and be more effectively treated. The center has gained national and international prominence with its studies on ethnic-associated cancer incidence and mortality rates and the role of dietary factors in cancer. There are 22 faculty who hold joint appointments with the School of Medicine or the School of Public Health and approximately 60 support staff.

6. Hawaii Down Syndrome Congress had established a database in 1985 and currently contains information on approximately 80 clients with Down Syndrome. The data includes demographic data including name, address, birthdate, family constellation; and minimal clinical data of major medical conditions. The database is used primarily as a membership list and as a mailing list to send out a newsletter.

7. Hemophilia Foundation maintains a limited database which includes the name and address of approximately 65 Hawaii residents. The database is used for mailing of information to individuals in the database.

8. Muscular Dystrophy Association maintains an automated data system with data for approximately 370 patients. The data includes demographic data, data collected at entry into the service and data related to the provision of clinical services by the association.

9. Medical Genetic Services data system was developed in 1977 and since that time has collected data on approximately 7,000 families. The data includes demographic data including ethnicity; elements of the history and

physical examinations, family history and pedigrees; and relevant medical data.

10. Hawaii Hereditary Hemoglobinopathy Project - Medical Genetic Services was developed in 1985 and currently has data on approximately 6,000 patients linked to 2,000 families. The data includes demographic data including ethnicity and geographic origin of the families; and relevant medical data related to the anemia.

11. Cytogenetics Laboratory (Kapiolani Medical Centers) database was established in January, 1993. The system was developed and is maintained by the Molecular and Clinical Cytogenetics Laboratory. The data base contains data on approximately 1,200 patients which includes patient specific demographic data and test results from chromosomal and DNA analysis.

12. Chorionic Villus Sampling Database was developed and is being maintained by the Fetal Diagnostic Center at Kapiolani Medical Centers. The data base contains data on approximately 350 patients who have undergone CV sampling which includes results of the procedures and pregnancy outcomes.

13. Triple Marker Database was developed by the Fetal Diagnostic Center at Kapiolani Medical Centers in May 1993. Data on approximately 100 patients who have had triplemark testing have been entered into the data base. The data includes names, medical chart number, referring physician, and results of the test. The data base is used for statistical purposes.

VI. RECOMMENDATIONS

Because the complexity of establishing and implementing a statewide system of genetic services, no one agency — public or private — has the capability and resources to address all of the problems alone. Therefore, it will take the coordinated and cooperative efforts of many organizations, agencies, and programs to achieve the recommendations.

The overall recommendation is for the Health Department to assume the leadership to convene a broad-based committee of public programs, community agencies, service providers, and academic programs to develop implementation plans for addressing the recommendations.

1. The Health Department's Children with Special Health Needs Branch should

consider consolidating all of the advisory committees into one overall advisory committee with ad hoc committees to address specific areas; and coordinate with other advisory committees that are not within the jurisdiction of the Health Department.

2. The Health Department's Children with Special Health Needs Branch should assume the leadership for the planning of a multi-agency collaborative effort to develop and implement a coordinated multi-media genetics awareness and educational campaign.

3. The Health Department's Newborn Screening Program in the Children with Special Health Needs Branch should continue to assess the feasibility and effectiveness of adding additional newborn screening tests.

4. The Health Department's Children with Special Health Needs Branch should assess the effectiveness of the Prenatal Screening Program and determine whether the activities should remain in the Department or be contracted through the purchase of service mechanism to a private agency.

5. The Health Department's Children with Special Health Needs Branch and the major medical facilities that provide services to women, children and families should examine alternative strategies for expanding the availability and improving the access to clinical genetic services across the state.

6. The Health Department should further explore the mechanism of contracting with the private sector some or all of the following services: public education, prenatal and newborn screening, birth defects monitoring, clinical genetics, and genetic counseling in order to assure the provision and continuation of these services.

7. The Health Department's Children with Special Health Needs Branch should explore the feasibility of consolidating the laboratory functions for prenatal and newborn screening into a centralized system for the purpose of maintaining quality control.

8. The Health Department's Children with Special Health Needs Branch should identify all of the persons responsible for the data systems related to genetic services and bring them together to discuss the feasibility of interfacing the data systems. This committee should assure that uniform data is being collected and that the analysis of the data is conducted in collaboration with the various genetics-related academic programs at the University of Hawaii.

9. There is a need to assess the feasibility and effectiveness of establishing a consistent funding base for the implementation of a screening program to identify individuals pre-conceptually who are at risk for hereditary anemias.

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